

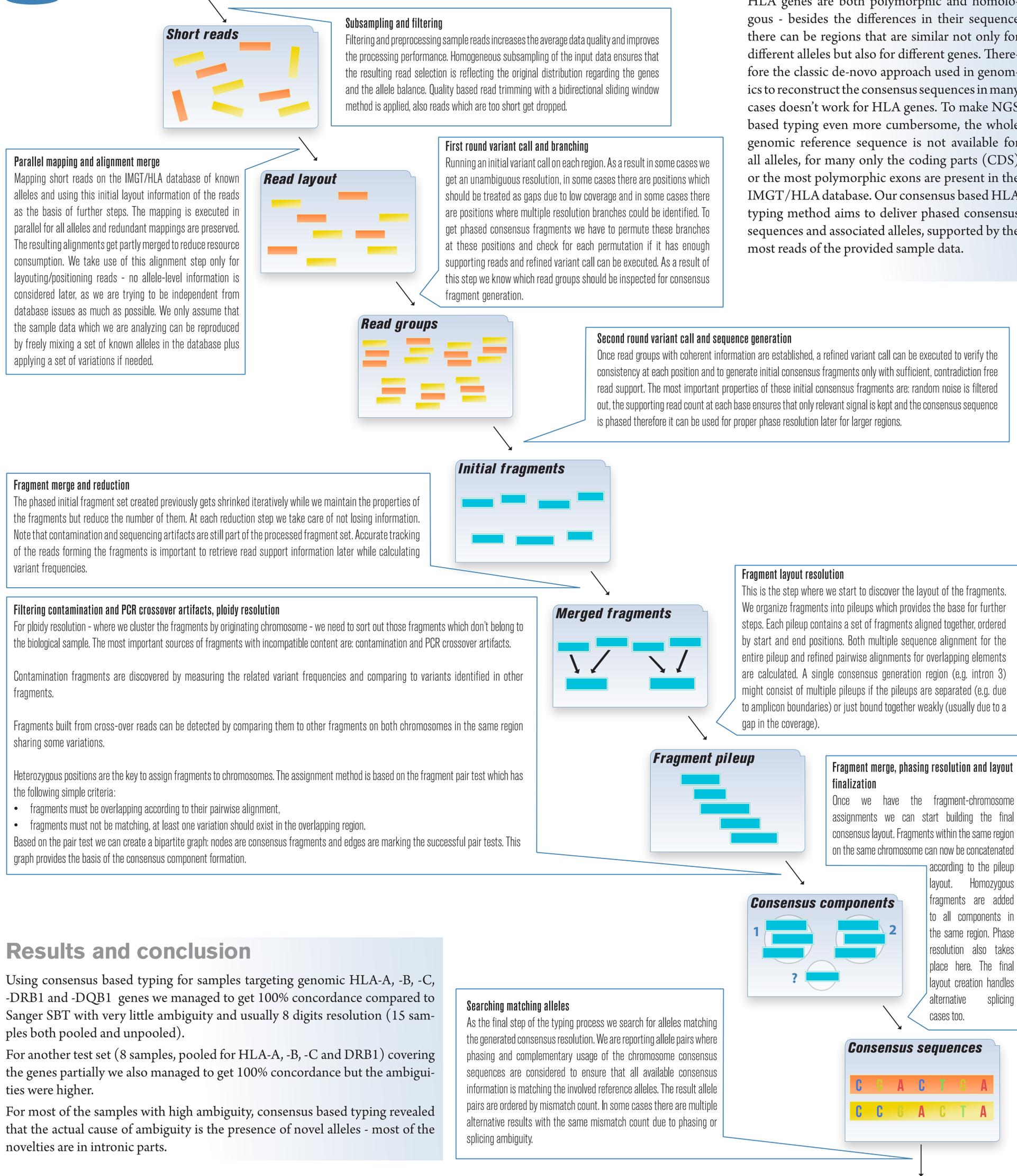
HIGH ACCURACY AND HIGH PRECISION HLA TYPING USING ILLUMINA READS:

TYPING ALGORITHM AND PERSPECTIVES ON NOVEL ALLELE DISCOVERY

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SAMPLE DATA



Introduction

HLA genes are both polymorphic and homologous - besides the differences in their sequence there can be regions that are similar not only for different alleles but also for different genes. Therefore the classic de-novo approach used in genomics to reconstruct the consensus sequences in many cases doesn't work for HLA genes. To make NGS based typing even more cumbersome, the whole genomic reference sequence is not available for all alleles, for many only the coding parts (CDS) or the most polymorphic exons are present in the IMGT/HLA database. Our consensus based HLA typing method aims to deliver phased consensus sequences and associated alleles, supported by the most reads of the provided sample data.

Results and conclusion

Using consensus based typing for samples targeting genomic HLA-A, -B, -C, -DRB1 and -DQB1 genes we managed to get 100% concordance compared to Sanger SBT with very little ambiguity and usually 8 digits resolution (15 samples both pooled and unpooled).

For another test set (8 samples, pooled for HLA-A, -B, -C and DRB1) covering the genes partially we also managed to get 100% concordance but the ambiguities were higher.

For most of the samples with high ambiguity, consensus based typing revealed that the actual cause of ambiguity is the presence of novel alleles - most of the novelties are in intronic parts.

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RESULT BROWSER